

Case Report

Bilateral dacryoceles associated with bilateral alacrimia with punctal and canalicular agenesis

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Abstract

To report a rare case of a 19 year old female presenting with bilateral dacryoceles and punctal and canalicular agenesis without epiphora. After clinical and radiological examination, the patient was found to have bilateral lacrimal gland agenesis as well. No other dental, otological or systemic abnormality suggestive of lacrimo-auricular-dento-digital syndrome was detected. She was managed with bilateral dacryocystectomy. Algorithm for managing such nonconventional lacrimal outflow dysgenesis based on presenting symptoms and co existing epiphora is proposed. Rationale of preferring lacrimal sac excision instead of dacryocystorhinostomy in this case is also discussed.

Keywords: Dacryocoele, Punctal agenesis, Lacrimal gland agenesis

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Introduction

Bilateral congenital dacryocoele without intranasal extension is a rare entity. Associated bilateral punctal and canalicular agenesis is rarer still. A MEDLINE search revealed only a single similar case report in the literature.¹ Lyon et al.² have listed 2 cases with bilateral punctal agenesis and medial canthal swelling as a part of their retrospective analysis of 57 patients with congenital punctal agenesis. Bilateral dacryocoele with punctal and canalicular agenesis and alacrimia form a part of lacrimo-auricular-dento-digital syndrome, also known as the Levy Hollister syndrome.³ To the best of our knowledge, this is the first case report of congenital bilateral dacryocoele with punctal, canalicular and lacrimal gland agenesis and no other finding of lacrimo-auricular-dento-digital syndrome was observed. Rare cases like these do not have fixed management protocols and the approach has to be customized as per patient's requirements.

Case

A 19 year old female presented with chief complaints of swelling appearing on the skin between the inner aspect of the eye and nose and watering of both eyes on and off, since birth. She also gave history of intermittent watering from both eyes since 5 years. Past ocular history revealed LASER refractive surgery (LASIK) done in both eyes 1 month ago.

The patient had cosmetic concerns regarding the bilateral swelling and did not find watering bothersome. There was no history of sticky discharge from the eye, nasal blockage or difficulty in breathing. She did not have any other eye complaints. Systemic history and birth history were not contributory.

Examination

Best corrected visual acuity in both eyes was 20/20. All four puncta were absent and there was a conjunctival fold

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bridging the punctal papilla and caruncle. A 3 cm by 2 cm circumscribed soft, cystic, nontender swelling was present in the right lacrimal sac area (Fig. 1). The swelling was located inferior to the medial canthal tendon and was displacing the medial canthus and medial end of lower lid margin upwards. A similar swelling measuring 2.5 by 1.7 cm was present on the left side. Skin overlying the swelling showed hyperpigmentation. The swelling was free from the overlying skin. There was no regurgitation on pressure over lacrimal sac area, or any lacrimal fistula. Tear secretion measured by Schirmers strips was normal. Anterior segment examination revealed a well opposed, healed LASIK flap. Rest of the posterior segment examination was within normal limits.

Based on these clinical findings, a diagnosis of bilateral congenital dacryoceles with punctal agenesis was made.

Direct nasal endoscopy of the right nose showed a normal inferior turbinate and inferior meatus without a nasolacrimal duct (NLD) opening. There was no intranasal extension of the lacrimal sac swelling. Rest of the meati and turbinates appeared normal. Similarly, the left nostril showed absence of NLD opening and no intranasal extension of sac swelling (Fig. 2).

Magnetic resonance imaging of orbit, paranasal sinuses and brain showed bilateral distended lacrimal sac (Fig. 3) with nasolacrimal duct atresia. Lacrimal gland was not visualized in any of the sections, bilaterally (Fig. 4). Apart from this, no other abnormality was detected.

Patient's primary concern was the prominent bilateral swelling. Bilateral dacryocystectomy was performed under local anesthesia. Intraoperatively, no canalicular remnants were identifiable.

Histopathology examination revealed distended lacrimal sac lined with pseudostratified columnar epithelium showing mucin secreting goblet cells (Fig. 5). Distended sac contents included desquamated cells and mucin. No inflammatory components were seen.

Post operative course was uneventful. Injection botulinum toxin to the lacrimal gland was offered for epiphora. However, patient did not find the epiphora bothersome and preferred to leave it alone. Patient is doing well at 6 months post operative follow up.

Discussion

The nasolacrimal outflow passage begins to develop at 6 weeks of gestational age and completes development by 7 months.⁴ It is derived from the surface ectoderm, which first

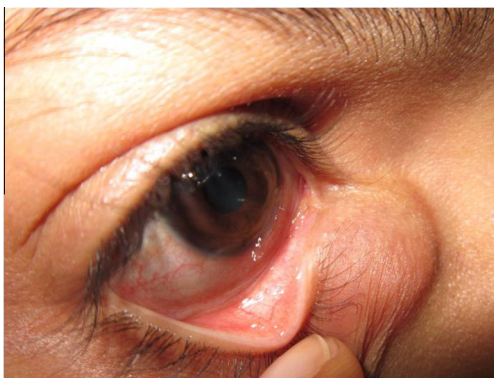


Figure 1. Lacrimal sac swelling with absent puncta.

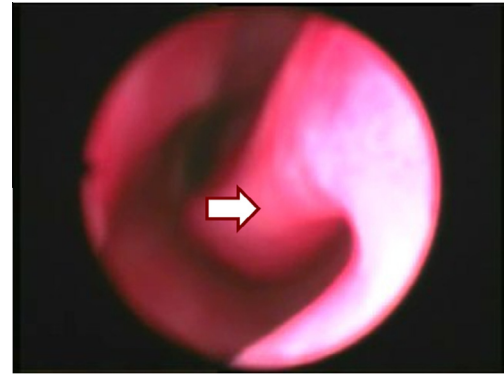


Figure 2. Nasal endoscopy-inferior turbinate.

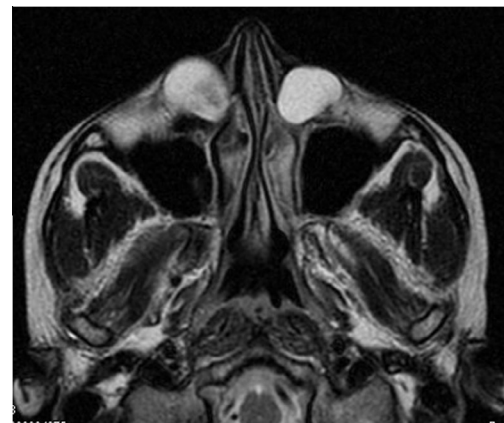


Figure 3. Bilateral dacryoceles on MR imaging.



Figure 4. No evidence of lacrimal gland on MR imaging.

forms a solid cord between the medial and lateral maxillary process. Later, this solid ectodermal cord canalizes by necrobiosis. This canalization begins at the sac and then proceeds cranially towards the canaliculi and caudally towards the nasolacrimal duct. When the lids separate at about 7 months gestation, the proximal end, that is, the puncta canalize. Shortly after, the distal most end, that is, the nasolacrimal duct canalizes. The various types of congenital lacrimal outflow abnormalities represent arrest of this development at corresponding stages.⁵ Congenital nasolacrimal duct obstruction (CNLDO), the commonest variety, represents an arrest at the last stage. If an arrest occurs one stage prior, even the puncta are affected, as evinced by the association of

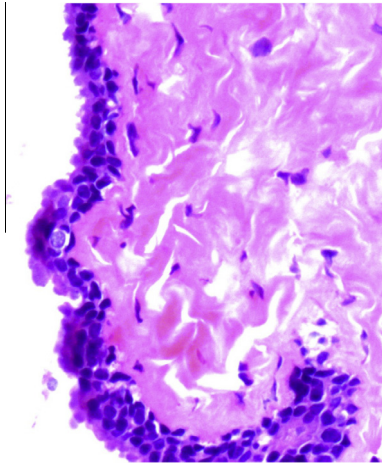


Figure 5. Lacrimal sac lining showing goblet cells on histopathology.

punctal anomalies in congenital nasolacrimal duct obstruction. Upto 13.6%⁶ of punctal anomalies have CNLDO. This case represents an arrest in the initial stages, when only the lacrimal sac canalized with a complete agenesis of lacrimal outflow system proximal as well as distal to the sac. This type of congenital bilateral dacryoceles is extremely rare.

Simultaneously, between 5 and 7 weeks the lacrimal gland develops from surface ectoderm. Our patient had bilateral lacrimal gland agenesis. Hence this condition represents a form of lacrimal apparatus dysgenesis due to embryological arrest in development.

Bilateral lacrimal sac swelling at birth could be due to congenital dacryocystocele, however, its pathogenesis is quite different. It represents a failure of canalization of valve of Hasner causing a distal obstruction. This leads to a proximal dilatation of the lacrimal sac and nasolacrimal duct. Distended lacrimal sac causes kinking of the common canaliculus, hence the sac becomes encysted. The puncta and canaliculi are generally not affected. Often, it has a nasal component and presents as neonatal respiratory distress in bilateral cases.

In a similar case previously reported^{1,2} the patient's presenting complaint was epiphora. He was successfully managed by endonasal dacryocystorhinostomy and silicone intubation. Upto 87%² of bilateral canalicular agenesis are associated with no underlying canalicular tissue. The current success rate for proximal canalicular reconstruction is modest (Dutton⁷ et al.), and bilateral congenital punctal atresia with canalicular agenesis represents a subset where in surgical outcomes are not encouraging. Despite having congenital punctal agenesis, our patient never had bothersome epiphora. The presence of co existing lacrimal gland agenesis is postulated to be the cause of lack of epiphora. Embryologically, the lacrimal gland and the lacrimal outflow pathway develop from the surface ectoderm between 5 and 7 weeks. Hence this condition represents a part of a wide spectrum of embryological anomalies ranging from a 'conventional' CNLDO to a more extensive lacrimal gland and lacrimal outflow dysgenesis.

Also, considering all similar cases reported so far,^{1,2} this condition may or may not be associated with epiphora, reflecting an underlying presence or absence of coexisting lacrimal gland hyposecretion. Whether this hyposecretion is due to downregulation of lacrimal gland secretions or developmental anomaly is a potential area of further research. Bilateral dacryoceles with punctal and canalicular agenesis and alacrimia forms a part of lacrimo-auricular-dento-digital (LADD) syndrome, also known as the Levy Hollister syndrome. However, this patient did not have any other abnormality of the teeth, ear or limbs, that are commonly associated with this syndrome. Hence, this could represent a mild variant of the LADD syndrome, with only ocular manifestations. Based on the presence or absence of epiphora, we recommend an algorithm for managing such cases (Fig. 6).

Our patient presented with cosmetic concerns and without bothersome epiphora. Hence the treatment goals were altered and simple excision of the lacrimal sac was preferred. A similar approach has been advocated by Allen⁸ et al. The patient was offered injection botulinum toxin in the lacrimal gland in case of bothersome epiphora.

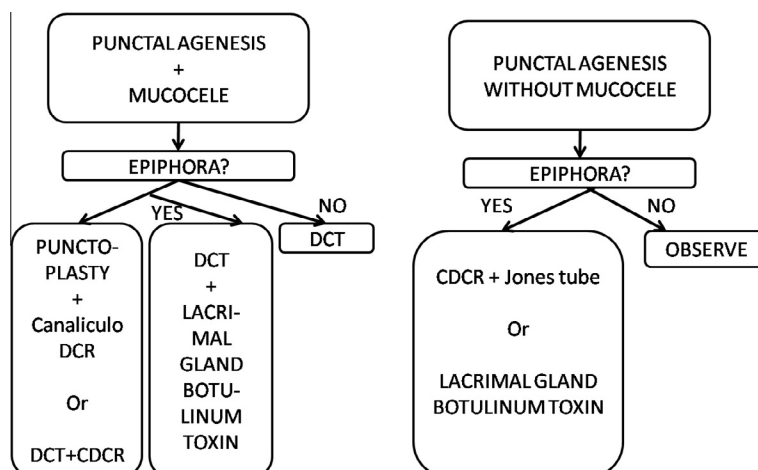


Figure 6. Algorithm for approach to punctal agenesis.

Conclusion

Bilateral congenital dacryoceles with punctal atresia and lacrimal gland agenesis represents a rare form of lacrimal outflow dysgenesis. This may represent a mild, ocular variant of the Levi Hollister syndrome, without any other system involvement. There are no set guidelines for its management. Epiphora may be absent due to coexisting lacrimal gland abnormality. Treatment approach needs to be customized as per patient's complaints. In case of absent epiphora, associated lacrimal gland abnormalities and agenesis should be looked into.

Conflict of interest

The authors declared that there is no conflict of interest.

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References

1. Ong CA, Prapegeran N, Sharad G, Luna D. Bilateral lacrimal sac mucocele with punctal and canalicular atresia. *Med J Malaysia* 2005;**6**(5):660–2.
2. Lyons CJ, Rosser PM, Welham RA. The management of punctal agenesis. *Ophthalmology* 1993;**100**(12):1851–5.
3. Santo RO, Golbert MB, Akaishi PM, Cruz AA, Cintra MB. Giant dacryocystocele and congenital alacrimia in lacrimo-auriculo-dento-digital syndrome. *Ophthal Plast Rec Surg* 2013;**29**(3):e67–8.
4. Burkat CM, Lucarelli MJ. Anatomy of the lacrimal system. In: Cohen AJ, Mercandetti M, Brazzo BG, editors. *The lacrimal systems. Diagnosis, management and surgery*. Springer; 2006. p. 3–4.
5. Yuen SA, Oley C, Sullivan TJ. Lacrimal outflow dysgenesis. *Ophthalmology* 2004;**111**(9):1782–90.
6. Ali MJ, Mohapatra S, Mulay K, Naik MN, Honavar SG. Incomplete punctal canalisation: the external and internal punctal membranes. Outcomes of membranotomy and adjunctive procedures. *Br J Ophthalmol* 2013;**97**(1):92–5.
7. Dutton JJ, Holck DE. Holmium laser canaliculoplasty. *Ophthal Plast Rec Surg* 1996;**12**(3):211–7.
8. Allen RC, Nerad JA. Re: "surgical treatment of familial dacryocystocele and lacrimal punctal agenesis". *Ophthal Plast Rec Surg* 2010;**26**(1):67.